

Susac syndrome presenting with acute hemibody paraesthesia

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Abstract

Susac syndrome is an orphan disease characterised by encephalopathy, branch retinal artery occlusion and sensorineural hearing loss. As the clinical triad is rarely present at symptom onset, it is often initially misdiagnosed and appropriate treatment is often delayed. Herewith, we report a case of Susac syndrome in a 47-year-old man presenting with acute hemisensory loss and highlight the challenges of early diagnosis, particularly relevant in the era of hyperacute stroke management.

Keywords

Acute stroke syndromes, behavioural changes and stroke, stroke in children and the young

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Introduction

We report a case of Susac syndrome in a 47-year-old man presenting with an acute hemisensory loss that could have been attributed to a lacunar stroke.

Case report

A 47-year-old right-handed male patient suffered an acute left hemibody paraesthesia on the way to work. He did not present to hospital, but over the following days developed frontal headaches and vomiting for which he attended the emergency department but was discharged following a normal CT head scan. Two months after his original presentation, the patient was found to be disorientated and highly distractible by his family and re-admitted to hospital. His clinical condition progressed to include more severe frontal headache, vomiting, worsening confusion and ataxia over a four-week period. His family also reported difficulty with hearing during this time. There was no history of fever, seizures or facial or limb weakness. Examination on the current admission revealed normal tone and power of the limbs but generalised symmetrical hyperreflexia, with bilateral Hoffman's responses and extensor plantar reflexes. Corrected visual acuities were 6/18 in the right and 6/12–1 on the left, with no relative afferent pupillary defects. He was initially treated with intravenous aciclovir for a suspected viral

encephalitis. Hepatitis A, B, and C, HIV, mycoplasma serology, Aquaporin 4 antibodies and voltage-gated potassium channel and N-Methyl-D-aspartate (NMDA)-receptor antibodies were all negative. Cerebrospinal fluid examination was acellular with normal glucose but revealed an opening pressure of 35 cmH₂O and elevated protein at 2.8 g/dL. Viral PCR, culture and oligoclonal bands were negative. A magnetic resonance imaging (MRI) brain scan (12 days from symptom onset) revealed 'snowball' white matter lesions in both cerebral hemispheres mostly centred on the splenium of the corpus callosum (Figure 1(a)). Audiometry revealed severe bilateral sensorineural hearing loss, especially on the right (Figure 1(b)), but responses were inconsistent. Otoacoustic emissions were absent bilaterally. Retinal imaging and retinal fluorescein angiography (RFA) revealed a

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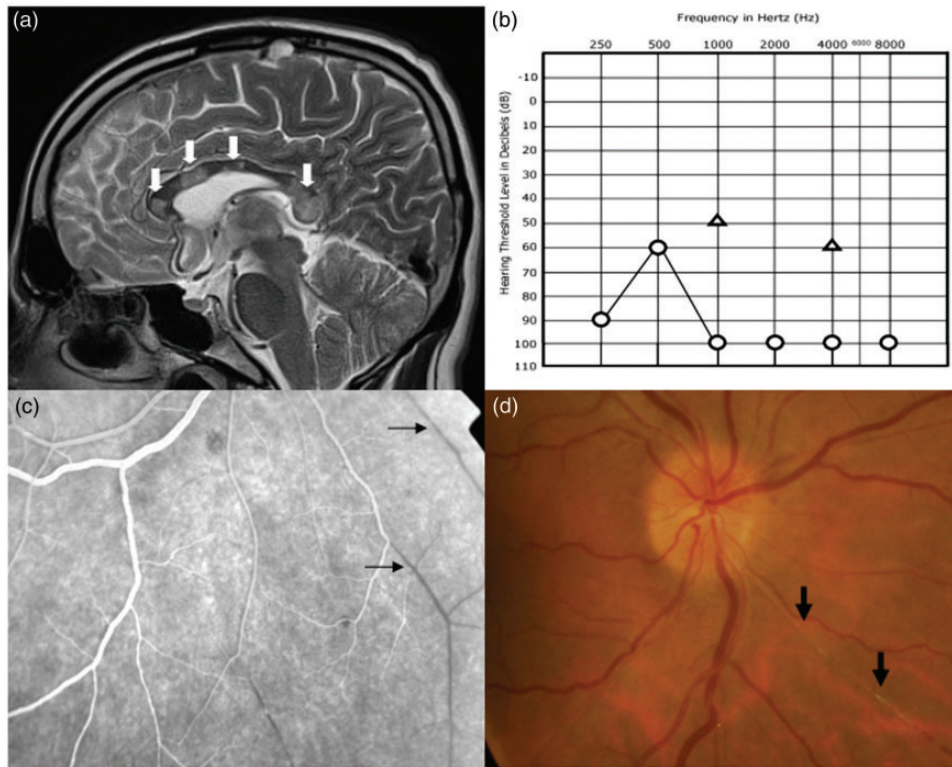


Figure 1. (a) T2 sagittal MRI brain showing high signal foci in the corpus callosum (white vertical arrows). Similar changes were also found in the subcortical white matter of the cerebral and cerebellar hemispheres bilaterally. (b) Right ear audiogram showing pan-frequency severe sensorineural hearing loss (circles) with relative sparing of bone conduction hearing (open triangles). (c) Fluorescein angiography showing filling defects in occluded branch retinal arteries (black horizontal arrows). There was evidence of multifocal fluorescence (not shown). (d) Retinal photography showing occluded branch retinal artery (black vertical arrows).

branch retinal artery occlusion in the right eye (Figure 1(c) and (d)), thus fulfilling the diagnostic criteria for Susac syndrome proposed by Kleffner et al.¹ Neuropsychometric testing two months into the illness was suggestive of profound global cognitive dysfunction with prominent frontal involvement. He was treated with pulsed methylprednisolone, followed by high-dose oral steroids. He remained mute, abulic, apathetic, and at times poorly responsive to visual and verbal stimuli. He had prominent gait ataxia. Given limited improvement, he underwent five cycles of plasma exchange followed by 1 g cyclophosphamide, initially fortnightly and then at monthly intervals, totalling 10 doses. At discharge four months after admission, he was fully alert, conversive and independently mobile but had severe hearing loss and marked underfunctioning in executive and attentional domains.

Discussion

We report a patient with Susac syndrome presenting with an acute hemibody paraesthesia which could

have initially been attributed to a contralateral lacunar stroke. Lacunar infarcts are small infarcts (2–20 mm in diameter) in the deep cerebral white matter, basal ganglia, or pons, presumed to result from the occlusion of a single small perforating artery. The lacunar syndromes are pure motor hemiparesis, ataxic hemiparesis, dysarthria/clumsy hand syndrome, pure sensory stroke, hemisensory loss of superficial sensation and sensorimotor stroke.² Whilst the initial presentation in our patient was compatible with a hemisensory lacunar stroke, the later development of a confusional state, behavioural change, a collateral history of hearing loss and abnormal MRI appearances within the corpus callosum all pointed towards Susac syndrome. Raised cerebrospinal fluid pressure as seen in our case has not been previously reported in Susac syndrome but may have been related to a non-specific central nervous system inflammatory response.

Susac syndrome is a retinocochleocerebral vasculopathy that describes the triad of encephalopathy, sensorineural hearing loss and branch retinal arterial occlusion.³ The pathophysiology of underlying Susac syndrome is yet to be fully elucidated but is

believed to involve immune-mediated injury to the vascular endothelium, leading to occlusion and subsequent ischaemic injury to the brain, cochlea and retina.⁴

Only 13% of patients present with the classic triad of symptoms, but virtually all develop the full triad during the course of the disease. Gait ataxia (seen in 25% of cases), sensory disturbance (24%) and upper motor neuron signs (21%) as found in our patient are also relatively common. In addition, 80% of patients report migraine-like headache at disease onset,⁵ or even in the preceding prodromal months, as was apparent in our patient. Hearing loss is the most common neurological manifestation but may be easily missed by both the patient and therefore the clinician. Hearing loss may be a dramatic and disabling feature of the condition as it is most often irreversible. Patients may benefit from hearing aids or, in more severe cases of hearing loss, cochlear implants.⁶

The absence of the full clinical triad at disease onset perhaps explains the delays in initial diagnosis; prompt diagnosis is nevertheless crucial given the delicate nature of the brain, retina and cochlea and the need to rapidly suppress the disease to avoid devastating damage.⁷ Diagnosis based on clinical symptoms alone can be difficult, particularly in encephalopathic or confused patients. Cerebral MRI, audiometry and RFA – with arterial wall hyperfluorescence distant to the branch retinal artery occlusion⁸ – are key investigations to make the diagnosis.⁹ Audiometry typically shows low to middle frequency hearing deficits.⁵ MRI typically reveals multifocal T2-hyperintense white matter lesions, often in the corpus callosum and periventricular areas,¹ explaining the confusional state and prominent frontal cognitive dysfunction.

Our patient required aggressive immunotherapy with cytotoxic medication owing to treatment failure with immunoglobulin and plasma exchange. This case expands the clinical spectrum of hemisensory loss in lacunar stroke mimics. We further highlight the importance of suspecting Susac syndrome even in the absence of the typical clinical triad. This is particularly salient in the current era of hyperacute stroke treatment where patients could be inappropriately treated for stroke.

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Contributorship

PC and DK researched literature. All authors conceived the report. PC wrote the first draft of the article. All authors reviewed and edited the article and approved the final version of the article.

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Ethical approval

Our institution does not require ethical approval for reporting individual cases or case series. Written informed patient consent was obtained for publication.


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References

1. Kleffner I, Dörr J, Ringelstein M, et al. Diagnostic criteria for Susac syndrome. *J Neurol Neurosurg Psychiatry* 2016; 87: 1287–1295.
2. Fisher CM. Lacunar strokes and infarcts: a review. *Neurology* 1982; 32: 871–876.
3. Susac JO, Hardman JM and Selhorst JB. Microangiopathy of the brain and retina. *Neurology* 1979; 29: 313–316.
4. Bitra RK and Eggenberger E. Review of Susac syndrome. *Curr Opin Ophthalmol* 2011; 22: 472–476.
5. Dorr J, Krautwald S, Wildemann B, et al. Characteristics of Susac syndrome: a review of all reported cases. *Nat Rev Neurol* 2013; 9: 307–316.
6. Lavinsky L, Scarton F, Lavinsky-Wolff M, et al. Successful cochlear implantation in a Susac syndrome patient. *Braz J Otorhinolaryngol* 2012; 78: 123.
7. Vodopivec I and Prasad S. Treatment of Susac syndrome. *Curr Treat Options Neurol* 2016; 18: 1–8.
8. Egan RA, Hills WL and Susac JO. Gass plaques and fluorescein leakage in Susac syndrome. *J Neurol Sci* 2010; 299: 97–100.
9. Rennebohm R, Susac JO, Egan RA, et al. Susac's syndrome – update. *J Neurol Sci* 2010; 299: 86–91.